What you should know when your Expanded AFP blood test result is

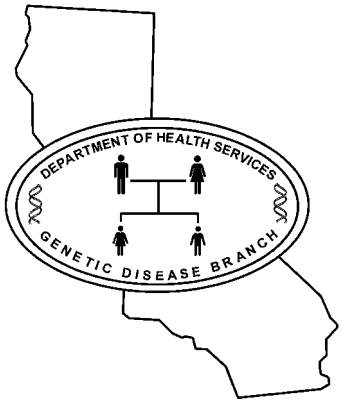
Screen Positive: Indicating an increased risk for trisomy 18

THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

POSITIVE: T18

the california expanded afp screening program

the california department of health services, genetic disease branch
850 marina bay parkway, f175, richmond, ca 94804
866-718-7915 *TOLL FREE*



As part of your prenatal care you had the Expanded AFP blood test. This screening test helps detect some birth defects such as trisomy 18, Down syndrome, neural tube defects and abdominal wall defects.

The blood test measured three substances in your blood: AFP (alpha-fetoprotein), HCG (human chorionic gonadotropin) and UE (unconjugated estriol). All of these substances are normally found in a woman's blood when she is pregnant.

Your test result was "screen positive" based on lower than expected amounts of AFP, HCG and UE in your blood. This indicates an increased risk (chance) that the fetus may have **trisomy 18**.

What could have caused your "screen positive" result?

Most of the time, the reason for this result is **NOT** a birth defect. Sometimes the low levels of AFP, HCG and UE occur without any known pregnancy problem.

To help determine *why your result was "screen positive"*, you will be offered diagnostic follow-up services at a State-approved Prenatal Diagnosis Center. There is no additional charge for approved services.

What can diagnostic tests show?

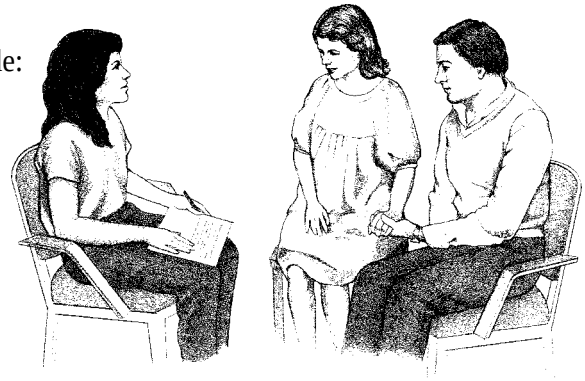
Certain birth defects, such as trisomy 18, can be found by examining the chromosomes of the fetus (unborn baby). Chromosomes are packages of genetic information found in every cell of the body. Birth defects can occur when the fetus has too few or too many chromosomes, or when there is a defective chromosome.

Most women with "screen positive" results will have normal follow-up tests and healthy babies.

What are the follow-up services at a State-approved Prenatal Diagnosis Center?

These follow-up services include:

- genetic counseling
- ultrasound
- amniocentesis



A woman may refuse any of these follow-up services at any time.

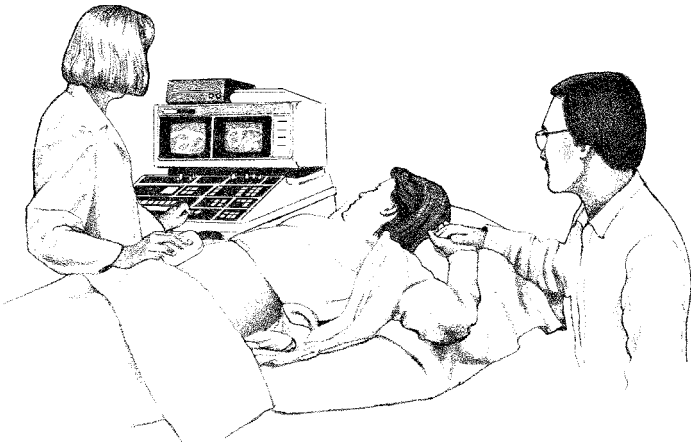
Genetic counseling:

A genetic counselor or doctor will discuss your blood test result and what it could mean. Your family's health history will be reviewed. Counseling will give you information to help you make decisions about the follow-up tests. Be sure to talk with the counselor or doctor about any questions or concerns you may have.

Ultrasound:

This test is also called a sonogram. Sound waves are used to make a picture of the fetus. This picture is seen on a special TV screen.

Ultrasound shows the age of the fetus and how many fetuses there are. It also may show whether there is a visible abnormality in the fetus or the uterus.



Amniocentesis is offered . . . .

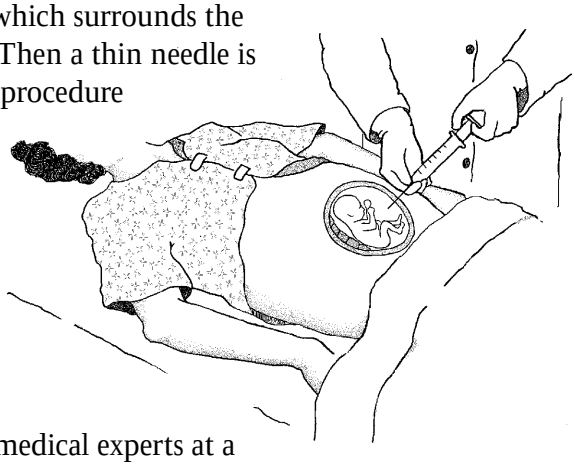
- ***if ultrasound does not explain the "screen positive" blood test result,***

or

- ***if ultrasound finds a problem which needs clarification.***

Amniocentesis:

This test involves removing a small amount of the amniotic fluid which surrounds the fetus. First, an ultrasound picture locates the fetus and the fluid. Then a thin needle is used to remove a small amount of the fluid from the uterus. This procedure sometimes causes brief discomfort. The amniotic fluid contains fetal cells. The chromosomes in these cells are counted and examined.



Amniocentesis detects almost all the birth defects caused by too many or too few chromosomes in the fetal cells. Trisomy 18 is an example of a birth defect caused by too many chromosomes.

The results of amniocentesis are usually ready in two weeks.

Amniocentesis is considered a safe procedure when performed by medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage following amniocentesis is less than 1%. Problems such as bleeding or infection are rare.

Sometimes not all of the follow-up tests are needed.
Genetic counseling will explain which tests will be useful.

Most amniocentesis results are normal.
Sometimes, however, a birth defect such as trisomy 18 is diagnosed.

Here are some facts about trisomy 18:

Trisomy 18 is caused by an extra chromosome #18. Infants born with this birth defect have severe mental retardation and very serious health problems. They usually die before birth or in early infancy. Trisomy 18 occurs in about 3 out of every 10,000 births in the United States.

If the fetus does have trisomy 18, will the follow-up tests detect it?

Amniocentesis detects almost 100% of the cases of trisomy 18 and other chromosome abnormalities.

What if the follow-up tests show that the fetus has a birth defect?

If a birth defect is found, a doctor or genetic counselor will give the woman or couple information about the defect and how it may affect the fetus. Available treatments and options for continuing or ending the pregnancy will be discussed.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

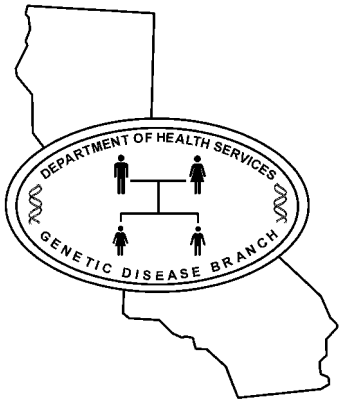
Please remember:

Most women who have had a "screen positive" test result will have normal healthy babies. However, in those pregnancies with a serious birth defect, early detection allows parents to explore their options and make early decisions.

The goal of the Department is to provide high quality, low cost services to all Californians. If you have questions, comments or suggestions about services received through this program please let us know.

Write to:

george c. cunningham, md
the california department of health services
genetic disease branch
850 marina bay parkway, f175
richmond ca 94804



1/2004